DEVELOPING THIS CHECKLIST

These recommendations have been developed according to the process recommended by the National Health and Medical Research Council of Australia.

A staged method was employed:

- comprehensive literature search to identify relevant information
- consultation with parents and caregivers on their experiences of poor growth, calorie intake and feeding difficulties
- preparation of a draft set of recommendations
- appointment of an international and multidisciplinary panel of expert clinicians who reviewed successive drafts of the recommendations until agreement was reached.

Expert Panel

Members of the expert panel who reviewed the earlier drafts are listed below and we thank them for their contribution.

Sue Abraham PhD, Albert Einstein College of Medicine, Montefiore Medical Center, Bronx, New York, USA;

Bruria Ben Zeev MD, Pediatric Neurology Institute, The Edmond & Lily Safra Children's Hospital, the Chaim Sheba Medical Center, Tel Hashomer, Israel;

Elana Bern MD, MPH Boston Children's Hospital, Boston, Massachusetts, USA;

Sudge Budden MD, Pediatric Development and Rehabilitation, Legacy Emanuel Children's Hospital, Portland, Oregon, USA;

Hilary Cass FRCPCH, Neuroscience Unit, Great Ormond Street Hospital for Children & Institute of Child Health, London, United Kingdom;

Carmelo Cuffari MD, The John Hopkins Hospital, Baltimore, Maryland, USA;

Carolyn Ellaway MBBS PhD FRACP CGHGSA, Western Sydney Genetics Program, The Children's Hospital at Westmead, Sydney, New South Wales, Australia;

John Fortunato Jr MD, Wake Forest Baptist Medical Center, Winston-Salem, North Carolina, USA;

Michael Freilinger MD, Department of Pediatrics and Adolescent Medicine, Medical University of Vienna, Vienna, Austria;

Suzanne Geerts RD MS, Civitan International Research Centre, University of Alabama, Birmingham, Alabama, USA;

Richard Haas MD, Department of Neurosciences, University of California San Diego, La Jolla, California, USA;

Peter Humphreys MD FRCP, Division of Neurology, Children's Hospital of Eastern Ontario, Ottawa, Ontario, Canada;

Mary Jones MD, Katie's Clinic for Rett Syndrome, Children's Hospital & Research Center, Oakland, California, USA;

Omar Khwaja MD PhD, Boston Children's Hospital, Boston, Massachusetts, USA;

Jane Lane RN BSN, Civitan International Research Centre, University of Alabama, Birmingham, Alabama, USA;

Ted O'Loughlin FRACP, Department of Gastroenterology, The Children's Hospital at Westmead, Sydney, New South Wales, Australia;

Alan Percy MD, Department of Pediatrics and Neurology, University of Alabama, Birmingham, Alabama, USA;

Mercè Pineda MD PhD, Department of Neuropediatrics, Hospital Sant Joan de Déu, Barcelona, Spain;

Carolyn Schanen MD PhD, Human Genetics Research, A.I. duPont Hospital for Children, Nemours Biomedical Research, Wilmington, Delaware, USA;

Catherine Senez BSc, Université Paris Diderot, Paris, France;

Steven Skinner MD, Greenwood Genetic Center, Greenwood, South Carolina, USA;

Eric Smeets MD PhD, Department of Clinical Genetics, Academic Hospital Maastricht, Maastricht, The Netherlands;

Sue Thompson BSc AdvAPD, Genetic Metabolic Disorders Service, The Children's Hospital at Westmead, Sydney, New South Wales, Australia;

Batya Weiss MD, Division of Pediatric Gastroenterology and Nutrition, The Edmond & Lily Safra Children's Hospital, the Chaim Sheba Medical Center, Tel Hashomer, Israel;

Helena Wandin PhD student, Department of Public Health and Caring Sciences, Research in Disability and Rehabilitation, Uppsala Universitet, Uppsala, Sweden;

Ingegerd Witt-Engerstrom MD PhD, Swedish Rett Center, Froson, Sweden.

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This checklist is based on the following:
Leonard H, Ravikumara M, Baikie G, Naseem N, Ellaway C, Percy A, Abraham S, Geerts S, Lane J, Jones M, Bathgate K, Downs J. *Assessment and management of nutrition and growth in Rett syndrome*. Journal of Pediatric Gastroenterology & Nutrition, 2013; 57:451-460.

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Authors of checklist

Jenny Downs, BAppSci MSc PhD
Helen Leonard, MBChB MPH
Deirdre Croft, BA DipCrim
Madhur Ravikumara, MBBS MRCPCH FRACP
Carolyn Ellaway, MBBS PhD FRACP CGHGSA
Sue Thompson, BSc AdvAPD
Alan Percy, MD FAAN FAAP
Gordon Baikie, MD FRACP

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Growth and nutrition in Rett syndrome

Checklist for clinicians on assessment and management

Rett syndrome is a rare neuro-developmental disorder that occurs almost exclusively in females. The prevalence rate is approximately 1:9000 of the female population.

Rett syndrome is caused by mutations in the methyl-CpG-binding protein 2 (*MECP2*) gene which affects the nerves in the central nervous system. This affects many systems of the body:

- the nervous system (cognitive impairment, communication difficulties, and epilepsy)
- the respiratory system (hyperventilation and breath holding)
- the musculo-skeletal system (altered muscle tone, contractures, low bone density and scoliosis)
- the gastrointestinal system (gastro-oesophageal reflux, gastrointestinal dysmotility, abdominal distension, constipation and oromotor dysfunction).

This leaflet is part of a series of publications developed by the Telethon Institute for Child Health Research in Western Australia to support better clinical assessment and management of health conditions commonly associated with Rett syndrome.

The content reflects issues cited by parent carers as important to them in responding to their daughter's growth and nutritional needs.

Other publications currently available in this series focus on clinical assessment and management of:

- *scoliosis in Rett syndrome*
- *nutrition and growth in Rett syndrome.*

A more comprehensive booklet on nutritional and digestive health in Rett syndrome has also been developed for families and carers and is available from ***rett.childhealthresearch.org.au***

Ensuring that females with Rett syndrome have a healthy nutritional intake is important to promote optimal bodily functioning.

The assessment and management of growth and nutrition in Rett syndrome needs to take account of:

- the likelihood of feeding difficulties (problems with chewing, swallowing and risk of aspiration)
- the likelihood of gastro-intestinal problems (reflux, abdominal bloating and constipation)
- possible increased energy requirements that may result from the susceptibility to breath holding and hyperventilation.

ASSESSMENT

It is recommended that a clinical assessment of growth and nutrition be conducted:

- approximately every six months from infancy to 12 years of age
- once a year throughout adolescence and adulthood.

Anthropometric Measures

- height, weight and BMI should be monitored regularly
- a standard growth chart can be used to plot anthropometric measures.
- growth charts for Rett syndrome are also now available (Tarquinio et al, 2012).
- upper arm circumference and/or triceps skin fold thickness may be used to assess nutritional status.

Recommendations for measuring height:

- use a stadiometer if the girl or woman with Rett syndrome is able to stand straight
- supine length may be measured for younger children (ie those less than two years)
- measure length of lower leg cross referenced against algorithmic base lines if musculoskeletal abnormality is present (Stevenson 1995).

Investigations

Biochemical tests conducted as part of the nutritional assessment should include testing levels for:

- full blood count, ferritin and vitamin B12
- urea, creatinine, and electrolytes, albumin/protein,
- Vitamin D, Calcium, phosphate and alkaline phosphatase.

Nutritional assessment

Nutritional assessment should include:

- caregiver reports of food intake, type and variety of foods, texture preferences and tolerance, and any supplements
- a 24 hour diet recall of food intake.

Feeding abilities

Functional capacities for self feeding, chewing and swallowing can be assessed by:

- detailed history from carers
- direct observation
- video of meal time
- video fluoroscopy.

A speech therapist or other appropriate allied health professional may be consulted for expert assessment of feeding, chewing and swallowing function. The Schedule for Oral Motor Assessment (SOMA) may be used to assess feeding in girls and women with Rett syndrome (Reilly et al 1995).

Feeding difficulties

Problems with the teeth and mouth may interfere with oral intake and should be carefully checked and a referral to a dentist made when appropriate.

Signs of feeding difficulties are indicated by signs of coughing, choking, gagging and/or crying. The time taken to feed is important and may be an indicator of carer stress if protracted.

Assessment should include the caregiver's report of appetite; length of typical meal time; drooling; spilling of food and drinks; regurgitation, bloating and/or constipation.

Assessment should explore the possibilities for reducing feeding difficulties through:

- physical positioning and postural supports
- special equipment and utensil use
- prompting and socialising during mealtimes
- modification of food consistency.

Video fluoroscopy is useful for the diagnosis of aspiration and to assess the need for texture modification.

MANAGEMENT

Whether the patient is under-weight or over-weight, nutritional intake should be tailored to individual requirements.

BMI at approximately the 25th centile can be considered a reasonable target for girls and women with Rett syndrome who are underweight. Height and wellness must be taken into account when setting clinical goals with attention not to exceed normal range.

Increasing calorie intake

The best indicator of calorie requirement can be obtained by comparing calorie intake to serial growth measures.

If the patient is under-weight, energy intake should be above the calorie requirments until a satisfactory weight has been attained.

To increase calories in the diet:

- supplement diet with energy-dense foods (as protein intake is usually adequate)
- provide frequent snacks of high calorie nutritional supplements.

NOTE:

- Gluten free and lactose free diets have not been associated with improved growth in Rett syndrome.

Measures to reduce feeding difficulties

Options may include:

- offering frequent small feeds throughout the day
- providing the food of choice
- modifying food textures and consistency
- physical positioning and postural support ensuring the jaw is supported and avoiding hyperextension
- special equipment and utensil use - trial different models of utensils, plates, cups and feeding bottles
- providing verbal and physical prompts for at least one meal a day to increase active participation in eating
- monitoring breathing patterns (feeding only when the rhythm of breathing is steady)
- trialling intensive therapy to optimise eating skills (research indicates that new skills can be mastered by some people with more severe feeding difficulties).

NOTE:

Girls and women with Rett syndrome may lose eating skills after an acute illness or surgery. Supportive treatment may be required until normal feeding returns.

If the person is living in out-of-home care, instructions on feeding techniques should be provided by an experienced nurse, dietician or speech therapist to all care staff concerned with feeding.

Enteral tube feeding

Enteral feeding options may improve the nutritional intake of females with Rett syndrome, as well as improve the quality of life of their caregivers.

Direct delivery of essential nutrients via the stomach, duodenum or jejunum may be considered when:

- there is a failure to gain weight despite efforts to increase calorie intake
- oromotor dysfunction is present with unsafe swallowing
- feeding times are protracted

Naso-gastric tube

NOTE: Insertion of a naso-gastric tube prior to insertion of a longer- term gastrostomy tube is usually a short term option but one which may help to:

- determine the potential for weight gain and growth improvement
- correct existing malnutrition (if caregivers are fearful of surgery or tube use and do not wish to commit to a permanent procedure prior to a trial).

Insertion of a naso-gastric tube may also assist to manage short term difficulties if:

- the feeding difficulty is likely to be temporary
- the person is acutely unwell.

Gastrostomy

As well as assuring adequate nutrition, insertion of a gastrostomy feeding tube can reduce feeding time. Girls or women can continue to enjoy meals, snacks and drinks if there is no aspiration risk, and the amount of feeds given by gastrostomy varies with oral intake. The gastrostomy can also be used to vent abdominal bloating which may help tolerance of oral and gastric feeds, and for administration of medications and fluid.

The procedure is most commonly performed via endoscopy and is called a PEG (Percutaneous Endoscopic Gastrostomy). Surgical insertion is sometimes necessary.

In cases of severe gastroesophageal reflux (GERD) that is not responsive to medical treatment, consideration should be given to performing a fundoplication procedure in addition to the gastrostomy.

In deciding whether or not to proceed with a gastrostomy, caregivers should be provided with information on the benefits and risks associated with the procedure. Practical and emotional support should be provided both before and after surgery.

Gastro-jejunostomy or PEG-J tube

Insertion of a feeding tube directly into the jejunum is recommended in cases of uncontrolled gastro oesophageal reflux or where gastrostomy feeds are not tolerated. A jejunostomy may also reduce the risk of aspiration where this is a risk. This can be done by a previous PEG (PEG-J) or placed surgically.

As well as the common complications associated with a gastrostomy, feeding through a jejunostomy tube may decrease the transit time of liquids leading to poor absorption of some nutrients. Feeds need to be given by continuous infusion.

Careful monitoring of *all* feeding tubes is required due to the possibility of the tube leaking, becoming dislodged or migrating. Other complications may include intestinal perforation, bleeding and an increased risk of reflux.

Clinical monitoring of enteral feeding procedures

Following insertion of a gastrostomy or gastro-jejunostomy, regular monitoring is required.

General condition

- assessing appearance and energy levels.

Blood chemistry

Any girl or woman who is malnourished or receiving at least 50% of daily nutritional needs via tube feeding should initially have a series of biochemical tests to establish a baseline, with a follow up six months later, and thereafter every 12 months testing for:

- full blood count, ferritin and liver function
- albumin/ protein, urea, electrolyte and creatinine
- calcium/phosphate/alkaline phosphatise and magnesium
- zinc and vitamins D, B12/folate.

Nutritional assessment

- monitoring calorie intake and energy requirements.

Fluid status

- monitoring all sources of fluid intake including enteral feeding, oral diet, water flushes of tubes, and any other oral intake of medication.

Gastro-intestinal function

- monitoring for vomiting, reflux, abdominal distension, pain and assessment of bowel function.

The feeding tube and stoma site

- checking position and functioning of feeding tube and health of stoma site.

Practical and emotional support

- assessing home management of equipment, and any other practical or emotional support needs.